

SEQUENCE LISTING

(1) GENERAL INFORMATION:

(i) APPLICANT:

- (A) NAME: BASF Aktiengesellschaft
- (B) STREET: Carl-Bosch-Strasse 38
- (C) CITY: Ludwigshafen
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(ii) TITLE OF APPLICATION: Method for diagnosing disorders by analysis of genes

(iii) NUMBER OF SEQUENCES: 2

(iv) COMPUTER READABLE FORM:

- (A) MEDIUM TYPE: Floppy disk
- (B) COMPUTER: IBM PC compatible
- (C) OPERATING SYSTEM: PC-DOS/MS-DOS
- (D) SOFTWARE: PatentIn Release #1.0, Version #1.25 (EPA)

(2) INFORMATION FOR SEQ ID NO: 1:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 1517 base pairs
- (B) TYPE: Nucleic acid
- (C) STRANDEDNESS: Double
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: cDNA for mRNA

(iii) HYPOTHETICAL: NO

(iv) ANTISENSE: NO

(vi) ORIGINAL SOURCE:

(A) ORGANISM: Homo sapiens

(ix) FEATURES:

(A) NAME/KEY: CDS

(B) LOCATION: 1..1024

(xi) SEQUENCE DESCRIPTION: SEQ ID NO: 1:

ATG	GGG	GAG	ATG	GAG	CAA	CTG	CGT	CAG	GAA	GCG	GAG	CAG	CTC	AAG	AAG		48
Met	Gly	Glu	Met	Glu	Gln	Leu	Arg	Gln	Glu	Ala	Glu	Gln	Leu	Lys	Lys		
1	5	10	15														
CAG	ATT	GCA	GAT	GCC	AGG	AAA	GCC	TGT	GCT	GAC	GTT	ACT	CTG	GCA	GAG		96
Gln	Ile	Ala	Asp	Ala	Arg	Lys	Ala	Cys	Ala	Asp	Val	Thr	Leu	Ala	Glu		
20	25	30	35	40	45												
CTG	GTG	TCT	GGC	CTA	GAG	GTG	GTG	GGA	CGA	GTC	CAG	ATG	CGG	ACG	CGG		144
Leu	Val	Ser	Gly	Leu	Glu	Val	Val	Gly	Arg	Val	Gln	Met	Arg	Thr	Arg		
35	40	45															
CGG	ACG	TTA	AGG	GGA	CAC	CTG	GCC	AAG	ATT	TAC	GCC	ATG	CAC	TGG	GCC		192
Arg	Thr	Leu	Arg	Gly	His	Leu	Ala	Lys	Ile	Tyr	Ala	Met	His	Trp	Ala		
50	55	60															
ACT	GAT	TCT	AAG	CTG	CTG	GTA	AGT	GCC	TCG	CAA	GAT	GGG	AAG	CTG	ATC		240
Thr	Asp	Ser	Lys	Leu	Leu	Ser	Ala	Ser	Gln	Asp	Gly	Lys	Leu	Ile			
65	70	75	80														
GTG	TGG	GAC	AGC	TAC	ACC	ACC	AAC	AAG	GTG	CAC	GCC	ATC	CCA	GTG	CGC		288
Val	Trp	Asp	Ser	Tyr	Thr	Thr	Asn	Lys	Val	His	Ala	Ile	Pro	Leu	Arg		
85	90	95															
TCC	TCC	TGG	GTC	ATG	ACC	TGT	GCC	TAT	GCC	CCA	TCA	GGG	AAC	TTT	GTG		336
Ser	Ser	Trp	Val	Met	Thr	Cys	Ala	Tyr	Ala	Pro	Ser	Gly	Asn	Phe	Val		

100	105	110	
GCA TGT GGG GGG CTG GAC AAC ATG TGT TCC ATC TAC AAC CTC AAA TCC			384
Ala Cys Gly Gly Leu Asp Asn Met Cys Ser Ile Tyr Asn Leu Lys Ser			
115	120	125	
CGT GAG GGC AAT GTC AAG GTC AGC CGG GAG CTT TCT GCT CAC ACA GGT			432
Arg Glu Gly Asn Val Lys Val Ser Arg Glu Leu Ser Ala His Thr Gly			
130	135	140	
TAT CTC TCC TGC TGC CGC TTC CTG GAT GAC AAC AAT ATT GTG ACC AGC			460
Tyr Leu Ser Cys Cys Arg Phe Leu Asp Asp Asn Asn Ile Val Thr Ser			
145	150	155	160
TCG GGG GAC ACC ACG TGT GCC TTG TGG GAC ATT GAG ACT GGG CAG CAG			528
Ser Gly Asp Thr Thr Cys Ala Leu Trp Asp Ile Glu Thr Gly Gln Gln			
165	170	175	
AAG ACT GTA TTT GTG GGA CAC ACG GGT GAC TGC ATG AGC CTG GCT GTG			576
Lys Thr Val Phe Val Gly His Thr Gly Asp Cys Met Ser Leu Ala Val			
180	185	190	
TCT CCT GAC TTC AAT CTC TTC ATT TCG GGG GCC TGT GAT GCC AGT GCC			624
Ser Pro Asp Phe Asn Leu Phe Ile Ser Gly Ala Cys Asp Ala Ser Ala			
195	200	205	
AAG CTC TGG GAT GTG CGA GAG GGG ACC TGC CGT CAG ACT TTC ACT GGC			672
Lys Leu Trp Asp Val Arg Glu Gly Thr Cys Arg Gln Thr Phe Thr Gly			
210	215	220	
CAC GAG TCG GAC ATC AAC GCC ATC TGT TTC CCC AAT GGA GAG GCC			720
His Glu Ser Asp Ile Asn Ala Ile Cys Phe Phe Pro Asn Gly Glu Ala			
225	230	235	240
ATC TGC ACG GGC TCG GAT GAC GCT TCC TGC CGC TTG TTT GAC CTG CGG			768
Ile Cys Thr Gly Ser Asp Asp Ala Ser Cys Arg Leu Phe Asp Leu Arg			
245	250	255	
GCA GAC CAG GAG CTG ATC TGC TTC TCC CAC GAG AGC ATC ATC TGC GGC			816
Ala Asp Gln Glu Leu Ile Cys Phe Ser His Glu Ser Ile Ile Cys Gly			
260	265	270	
ATC ACG TCT GTG GCC TTC TCC CTC AGT GGC CGC CTA CTA TTC GCT GGC			864
Ile Thr Ser Val Ala Phe Ser Leu Ser Gly Arg Leu Leu Phe Ala Gly			
275	280	285	
TAC GAC GAC TTC AAC TGC AAT GTC TGG GAC TCC ATG AAG TCT GAG CGT			912
Tyr Asp Asp Phe Asn Cys Asn Val Trp Asp Ser Met Lys Ser Glu Arg			
290	295	300	
GTG GGC ATC CTC TCT GGC CAC GAT AAC AGG GTG AGC TGC CTG GGA GTC			960
Val Gly Ile Leu Ser Gly His Asp Asn Arg Val Ser Cys Leu Gly Val			
305	310	315	320
ACA GCT GAC GGG ATG GCT GTG GCC ACA GGT TCC TGG GAC AGC TTC CTC			1008
Thr Ala Asp Gly Met Ala Val Ala Thr Gly Ser Trp Asp Ser Phe Leu			
325	330	335	
AAA ATC TGG AAC TGA G GAGGCTGGAG AAAGGGAAGT GGAAGGCAGT GAACACACTC			1064
Lys Ile Trp Asn *			
340			
AGCAGCCCCC TGCCCCGACCC CATCTCATTC AGGTGTTCTC TTCTATATTG CGGGTGCCAT			1124
TCCCCACTAAG CTTTCTCCTT TGAGGGCAGT GGGGAGCATG GGACTGTGCC TTTGGGAGGC			1184
AGCAGTCAGGG ACACAGGGC AAAGAACTGC CCCATCTCCT CCCATGGCCT TCCCTCCCCA			1244
CAGTCCTCAC AGCCTCTCCC TTAATGAGCA AGGACAAACCT GCCCCCTCCCC AGCCCTTTC			1304
AGGCCAGCA GACTTGAGTC TGAGGCCCA GGCCCTAGGA TTCCTCCCCC AGAGCCACTA			1364
CCTTTGTCCA GGCCCTGGGTG GTATAGGGCG TTTGGCCCTG TGACTATGGC TCTGGCACCA			1424
CTAGGGTCCT GGCCCTCTTC TTATTCTATGC TTTCTCCTTT TTCTACCTTT TTTCTCTCC			1484
TAAGACACCT GCAATAAAAGT GTAGCACCCCT GGT			1517

(2) INFORMATION FOR SEQ ID NO: 2:

(i) SEQUENCE CHARACTERISTICS:

(A) LENGTH: 341 amino acids

(3) TYPE: Amino acid

(D) TOPOLOGY: linear

(ii) MOLECULE TYPE: Protein

(xi) SEQUENCE DESCRIPTION: SEQ ID NO: 3

(X1) SEQUENCE DESCRIPTION: SEQ ID NO: 2:
 Met Gly Glu Met Glu Gln Leu Arg Gln Glu Ala Glu Gln Leu Lys Lys
 1 5 10 15
 Gln Ile Ala Asp Ala Arg Lys Ala Cys Ala Asp Val Thr Leu Ala Glu
 20 25 30
 Leu Val Ser Gly Leu Glu Val Val Gly Arg Val Gln Met Arg Thr Arg
 35 40 45
 Arg Thr Leu Arg Gly His Leu Ala Lys Ile Tyr Ala Met His Trp Ala
 50 55 60
 Thr Asp Ser Lys Leu Leu Val Ser Ala Ser Gln Asp Gly Lys Leu Ile
 65 70 75 80
 Val Trp Asp Ser Tyr Thr Thr Asn Dns Val His Ala Ile Pro Leu Arg
 85 90 95
 Ser Ser Trp Val Met Thr Cys Ala Tyr Ala Pro Ser Gly Asn Phe Val
 100 105 110
 Ala Cys Gly Gly Leu Asp Asn Met Cys Ser Ile Tyr Asn Leu Lys Ser
 115 120 125
 Arg Glu Gly Asn Val Lys Val Ser Arg Gln Leu Ser Ala His Thr Gly
 130 135 140
 Tyr Leu Ser Cys Cys Arg Phe Leu Asp Asp Asn Asn Ile Val Thr Ser
 145 150 155 160
 Ser Gly Asp Thr Thr Cys Ala Leu Trp Asp Ile Glu Thr Gly Gln Gln
 165 170 175
 Lys Thr Val Phe Val Gly His Thr Gly Asp Cys Met Ser Leu Ala Val
 180 185 190
 Ser Pro Asp Phe Asn Leu Phe Ile Ser Gly Ala Cys Asp Ala Ser Ala
 195 200 205
 Lys Leu Trp Asp Val Arg Glu Gly Thr Cys Arg Gln Thr Phe Thr Gly
 210 215 220
 His Glu Ser Asp Ile Asn Ala Ile Cys Phe Phe Pro Asn Gly Glu Ala
 225 230 235 240
 Ile Cys Thr Gly Ser Asp Asp Ala Ser Cys Arg Leu Phe Asp Leu Arg
 245 250 255
 Ala Asp Gln Glu Leu Ile Cys Phe Ser His Glu Ser Ile Ile Cys Gly
 260 265 270
 Ile Thr Ser Val Ala Phe Ser Leu Ser Gly Arg Leu Leu Phe Ala Gly
 275 280 285
 Tyr Asp Asp Phe Asn Cys Asn Val Trp Asp Ser Met Lys Ser Glu Arg
 290 295 300
 Val Gly Ile Leu Ser Gly His Asp Asn Arg Val Ser Cys Leu Gly Val
 305 310 315 320
 Thr Ala Asp Gly Met Ala Val Ala Thr Gly Ser Trp Asp Ser Phe Leu
 325 330 335
 Lys Ile Trp Asn *
 340

We claim:

1. The use of a genetic modification in the gene for human G protein $\beta 3$ subunit for the diagnosis of diseases.
- 5 2. The use of a genetic modification in the gene for human G protein $\beta 3$ subunit for establishing the risk of developing a disorder associated with G protein dysregulation.
- 10 3. The use as claimed in claim 2, wherein the genetic modification is in the codon for amino acid 275 in SEQ ID NO:1.
- 15 4. The use as claimed in claim 3, wherein there is substitution of cytosine by thymine in position 825 in SEQ ID NO:1.
- 5 5. The use as claimed in claim 2, wherein the disorder is a cardiovascular disease, a metabolic disturbance or an immunological disease.
- 20 6. The use as claimed in claim 2, wherein the disorder is hypertension.
- 25 7. A method for establishing a relative risk of developing disorders associated with G protein dysregulation for a subject, which comprises comparing the gene sequence for human G protein $\beta 3$ subunit of the subject with the gene sequence SEQ ID NO:1, and, in the event that a thymine (T) is present at position 825, assigning the subject an increased risk of disease.
- 30 8. A method as claimed in claim 7, wherein the comparison of genes is carried out by sequencing.
- 35 9. A method as claimed in claim 8, wherein a gene section which includes position 825 is amplified before the sequencing.
10. A method as claimed in claim 7, wherein the comparison of genes is carried out by hybridization.
- 40 11. A method as claimed in claim 7, wherein the comparison of genes is carried out by cleavage using restriction enzymes.
- 45 12. A method as claimed in claim 11, wherein the restriction enzyme Dsa I is used.

The use of a genetic modification in the gene for human G protein β_3 subunit for the diagnosis of diseases

5 Abstract

The present invention relates to the use of a genetic modification in the gene for human G protein β_3 subunit for the diagnosis of diseases.

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add b?

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add c?

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